



NATIONAL ACTION PLAN ON BREAST CANCER  
A Public/Private Partnership

W O R K I N G G R O U P

JANUARY 1999

THE HEREDITARY SUSCEPTIBILITY WORKING GROUP

One of the most exciting advances in breast cancer research within the last 10 years has been the discovery of two genes for breast cancer—BRCA1 and BRCA2. About 5 to 10 percent of all breast cancer patients carry a BRCA gene mutation. BRCA1 is thought to account for up to 50 percent of inherited breast cancers and up to 25 percent of breast cancer cases in women under the age of 30. Other inherited diseases, such as ataxia telangiectasia and Li-Fraumeni syndrome, also have been linked to an increased risk of breast cancer.

WHAT IS THE NAPBC?

The National Action Plan on Breast Cancer (NAPBC) was established in 1993 in response to a National Breast Cancer Coalition petition signed by 2.6 million people and presented to President Clinton. The petition called for a coordinated national strategy to combat breast cancer, the second leading cause of cancer deaths among American women.

The mission of the NAPBC is to stimulate rapid progress in eradicating breast cancer. The strategy for the Plan is to:

- *encourage new ideas and define unaddressed breast cancer priority areas.*
- *serve as a catalyst for national efforts to advance breast cancer knowledge, research, policy, and services.*
- *mobilize partnerships and coordinate actions among diverse public and private sector organizations and individuals.*

The work of the NAPBC is guided by a Steering Committee and Working Groups. The Steering Committee provides oversight for and ensures coordination across NAPBC initiatives. The Working Groups correspond to Plan priorities and identify, recommend, and oversee implementation of Plan activities. Plan implementation is coordinated by the U.S. Public Health Service's Office on Women's Health. Currently, there are five Working Groups. This fact sheet describes the Hereditary Susceptibility Working Group.

These findings are among the many exciting genetic discoveries that are both deepening our understanding of health and disease and transforming the practice of medicine. Discoveries about specific genetic mutations and their association with disease carry great promise for identifying people who may develop a disease in the future, for detecting disease early enough to begin effective and less invasive treatment, and ultimately to prevent the occurrence of the disease. At the same time, however, this rapid pace of scientific discovery poses major challenges. Society must develop policies and programs to respond to the clinical, ethical, legal, and psychosocial ramifications of identifying healthy individuals who may carry a cancer gene mutation.

These genetic discoveries pose challenging questions and immediate needs. How will we ensure that physicians and other health care providers are informed about new genetic discoveries and their clinical implications for early detection and treatment? How do we ensure that women who are tested for a breast cancer gene mutation are protected against job or insurance discrimination? Who should undergo genetic testing? What, if any, medical action should a woman take if a test reveals that she carries an error in a breast cancer gene? What are the implications for a woman's children?

The Hereditary Susceptibility Working Group of the National Action Plan on Breast Cancer (NAPBC) was formed to address these challenging issues. Its charge is to develop and implement a comprehensive plan to address the needs of individuals carrying breast cancer susceptibility genes and to recommend educational interventions for consumers, health care professionals, at-risk population groups, and policymakers in government and industry.

## **CURRENT ACTIVITIES AND ACCOMPLISHMENTS**

The recent explosion in genetic knowledge, the increasing availability of genetic information through commercial testing, and the intense media attention devoted to the issue have created an enormous need for education among health care providers, consumers, and others. The Working Group has moved rapidly on several fronts to fill this need.

### **Position Paper on Cancer Predisposition Testing**

The Hereditary Susceptibility Working Group developed a position paper, "Hereditary Susceptibility Testing for Breast Cancer," which addresses such issues as requiring cancer genetics education of health care providers, informed consent for individuals considering testing, and prevention of genetic discrimination. This document was published as a commentary to a statement from the American Society of Clinical Oncology in the *Journal of Clinical Oncology*. The paper can be viewed on the NAPBC Web site (<http://www.napbc.org>).

### **Curriculum Guidelines for Health Care Professionals**

In another effort to reach health care professionals, the Working Group developed educational curriculum guidelines. A workshop convened in April 1996 brought together scientists, health care providers, and breast cancer survivors and their families to discuss the need for and content of an educational curriculum on the implications of genetic testing for breast cancer susceptibility. A curriculum outline was then developed for use by health care professional groups to enable them to provide high-quality, customized education and training for their members on this complex and pressing issue. The outline was disseminated to nursing and medical schools and is available on the NAPBC Web site.

### **Consumer Education about Genetic Testing**

The Working Group also has focused on the need to educate consumers, through their health care providers, about genetic testing. The Working Group's educational videotape, *Genetic Testing for Cancer Risk: It's Your Choice*, addresses many of the pros and cons, as well as psychosocial issues, involved in genetic testing. This 15-minute video introduces the public—especially families with a history of breast and ovarian cancer—to the new technology of genetic testing for risk assessment. Information on ordering the video can be obtained on the NAPBC Web site or by calling (301) 294-5407.

### **Resources on Hereditary Susceptibility**

The Working Group has designed a searchable database, the Hereditary Susceptibility Materials Database, as a resource on hereditary susceptibility to breast cancer. The database contains materials developed by cancer agencies and organizations, as well as educational institutions, that are available to consumers and health care professionals. It will be placed on the NAPBC Web site in early 1999.

**WORKING GROUP ACCOMPLISHMENTS**

*Developed the position paper, "Hereditary Susceptibility Testing for Breast Cancer," published in the Journal of Clinical Oncology (14:1738-40, 1996).*



*Developed educational curriculum guidelines for health care professionals on genetic testing for breast cancer.*



*Produced and disseminated an educational video for consumers on genetic testing.*



*Convened the Consumer Leadership Summit: Preparing for the New Era of Genetics and Medicine.*



*Identified issues and developed recommendations on genetic discrimination in health insurance for use by Federal and State policymakers (published in Science 270:391-3, 1995).*



*Identified issues and developed recommendations on genetic information and the workplace (published in Science 275:1755-7, 1997).*



*Held a workshop to identify research priorities in hereditary breast cancer. The National Cancer Institute has incorporated many of these research recommendations into its program, the National Cancer Genetics Network.*



*Convened the Workshop on Privacy and Confidentiality in Genetics Research to develop principles that will prevent discrimination on the basis of an individual's genetic information.*



*Identified issues and developed recommendations related to the privacy of genetics research information.*

**Consumer Leadership Summit**

The Consumer Leadership Summit: Preparing for the New Era of Genetics and Medicine was held in December 1996 in collaboration with the National Breast Cancer Coalition. This conference was designed to promote education about genetic testing among various health advocacy and professional groups, to familiarize groups with the pros and cons of genetic testing, and to develop strategies for the dissemination of educational materials.

**Health Insurance Issues**

In the area of policy development, the Hereditary Susceptibility Working Group has led several efforts to examine issues of genetic discrimination by health and life insurance providers and in the workplace. In July 1995, the Working Group convened the Workshop on Genetic Information and Health Insurance. The workshop, cosponsored by the National Institutes of Health-Department of Energy Working Group on Ethical, Legal, and Social Issues of Human Genome Research (ELSI Working Group), was designed to provide a framework and establish recommendations for Federal and State policies to address the growing potential for genetic discrimination in health insurance coverage. The recommendations that emerged from the workshop were published in *Science* and are available on the NAPBC Web site.

**Employment Issues**

Also in collaboration with the ELSI Working Group, the Hereditary Susceptibility Working Group conducted the Workshop on Genetic Information and the Workplace in October 1996. This meeting addressed the adequacy of the Americans with Disabilities Act to protect against genetic discrimination, presented case studies of discrimination in insurance and employment, discussed Federal and State activities to prohibit discrimination, and explored issues related to the potential use and misuse of genetic information by employers. Recommendations for Federal and State policies that emerged from the workshop were published in *Science*, as well as posted on the NAPBC Web site.

In related efforts, the Working Group was invited to provide information to Federal and State policymakers on the legal and ethical issues surrounding the discovery of breast cancer susceptibility genes, providing a foundation for the establishment of policies to protect the privacy of genetic information in relation to health and life insurance coverage and employment.

### Research Gaps and Priorities

In addition to its work in the education and policy arenas, the Hereditary Susceptibility Working Group has focused attention on identifying gaps in current research. To identify these research gaps and to establish priorities for action, the Working Group held a workshop in September 1996 on the State-of-the-Science on Hereditary Breast Cancer. Speakers made presentations on BRCA1 function and biology, population and prevalence studies, gene-gene and gene-environment interactions, prevention, surveillance and treatment, and psychosocial research. Building on the foundation provided by the presentations, the participants met in small groups to identify and set priorities among emerging research needs. The Working Group has developed workshop proceedings, which were widely distributed to the scientific, health care, consumer, and policymaking communities. Many of the recommendations from the workshop have been incorporated into the National Cancer Institute's (NCI's) National Cancer Genetics Network. Workshop recommendations can be viewed on the NAPBC Web site.

### Workshop on Privacy and Confidentiality in Genetics Research

The goal of this workshop, which was conducted in September 1997, was to address privacy and confidentiality issues related to genetic information and to promote biomedical research. Prior to the workshop, a background paper was prepared and distributed to workshop participants to help them achieve specific goals. Recommendations resulting from the workshop will be published in early 1999.

### Providing Input into Related Initiatives

An important feature of these Working Group activities has been the partnerships established in their implementation, many of which have continued in further initiatives. For example, the Working Group has established ties with NCI's National Cancer Genetics Network. This group is developing a research network of health care professionals across the country who will be able to discuss issues of genetic testing with patients and other consumers. The Working Group is ensuring that the Network's initiatives acknowledge and incorporate consumer concerns about genetic testing, such as access, informed consent, privacy, and the need to weigh the pros and cons of testing.

#### FOR MORE INFORMATION

For more information about the Hereditary Susceptibility Working Group and its activities, please contact:

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#### FUTURE PLANS

Legislation and policy concerning hereditary susceptibility to breast cancer are rapidly changing areas that require continual examination. On February 19, 1999, the Working Group will convene a workshop, *Braddon v. Abbott*: Implications for Asymptomatic Genetic Conditions, to address the complex, unique, and potentially controversial questions regarding predictive genetics and breast cancer that were raised in a recent Supreme Court decision. In *Braddon v. Abbott*, the Supreme Court held that asymptomatic HIV infection is a disability under the Americans with Disabilities Act. Using breast cancer as a case study, the workshop will explore the implications of this decision for individuals with asymptomatic genetic conditions.